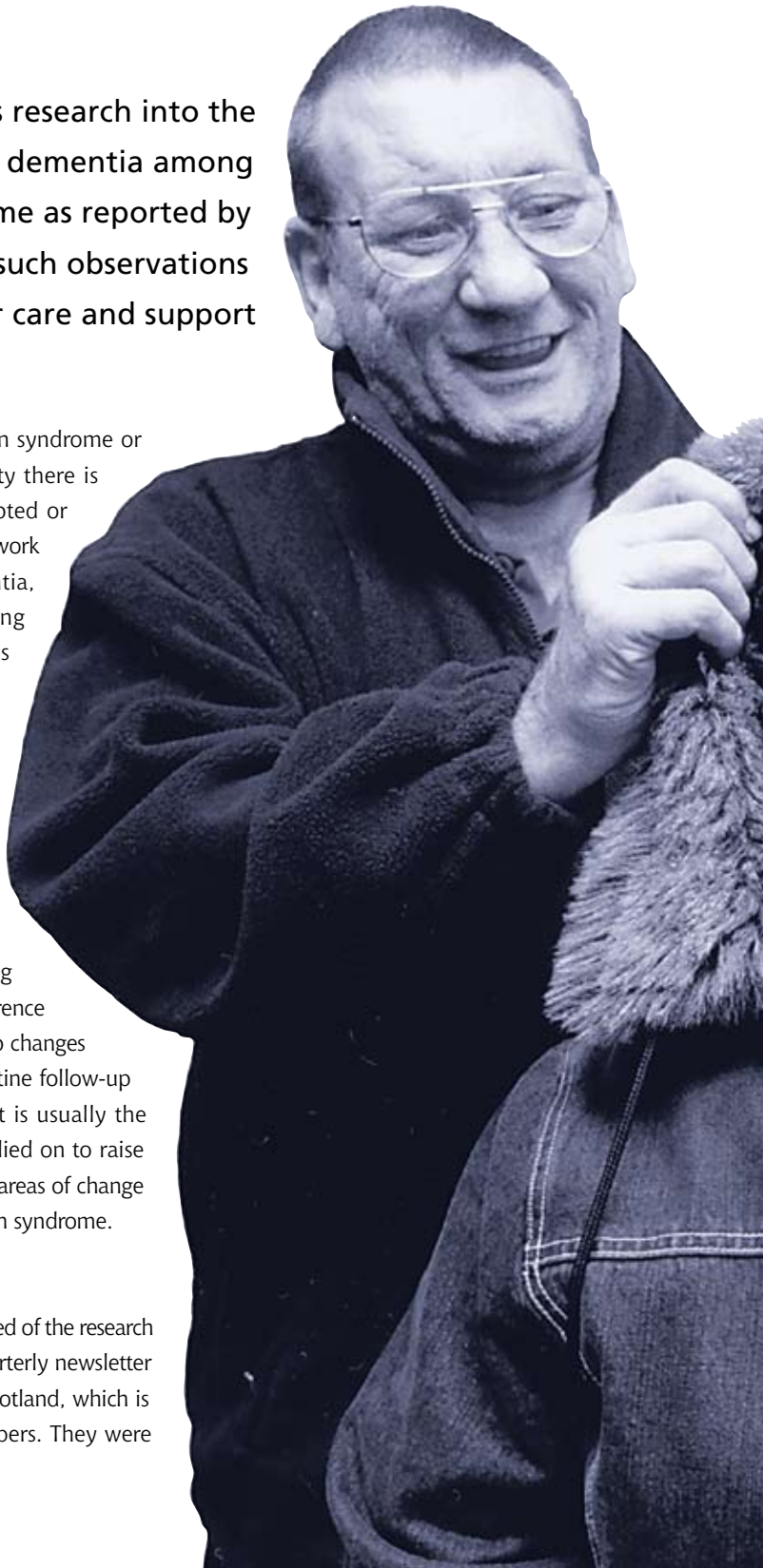
dementia and Down syndrome:

the diagnosis and support needed

Keywords
- Down syndrome
- dementia
- diagnosis

These keywords are based on the subject headings from the British Nursing Index. This article has been subject to a double-blind review.

Karen Watchman describes research into the early signs and symptoms of dementia among people with Down syndrome as reported by carers. She suggests that such observations can help shape later care and support



Recent research has suggested that the early symptoms and signs of dementia in people with Down syndrome may be different from those experienced by the general population (Ball *et al* 2006) and that the diagnosis of dementia is not routinely given to a person with Down syndrome (Wilkinson *et al* 2004). This article, following original research, suggests that early symptoms and signs of dementia reported by carers may be linked to understanding the type of dementia a person is experiencing. This understanding can be crucial to future care because using carers' reports of early symptoms of dementia may help establish an accurate diagnosis and guide appropriate interventions.

Background

In the wider population without Down syndrome a detailed individual history and tests of memory function can usually assist in establishing a diagnosis of dementia, in conjunction with physical and neurological assessments including brain imaging. Results, while not always conclusive, have a high validity, although the underlying pathology of dementia may not be established without eventual post-mortem.

For adults with Down syndrome or other learning disability there is still not a widely accepted or consistently used framework for diagnosing dementia, despite a long-standing awareness that this group is at a greater risk of dementia at a younger age (Janicki *et al* 2000). Baseline assessments, usually based on psychological test instruments and questionnaires, are increasingly being used to establish a reference from which to follow up changes (Dodd *et al* 2001). Routine follow-up is not consistent and it is usually the carer who is initially relied on to raise concerns about specific areas of change in the person with Down syndrome.

Method

Participants were informed of the research project through the quarterly newsletter of Down's Syndrome Scotland, which is distributed to all members. They were

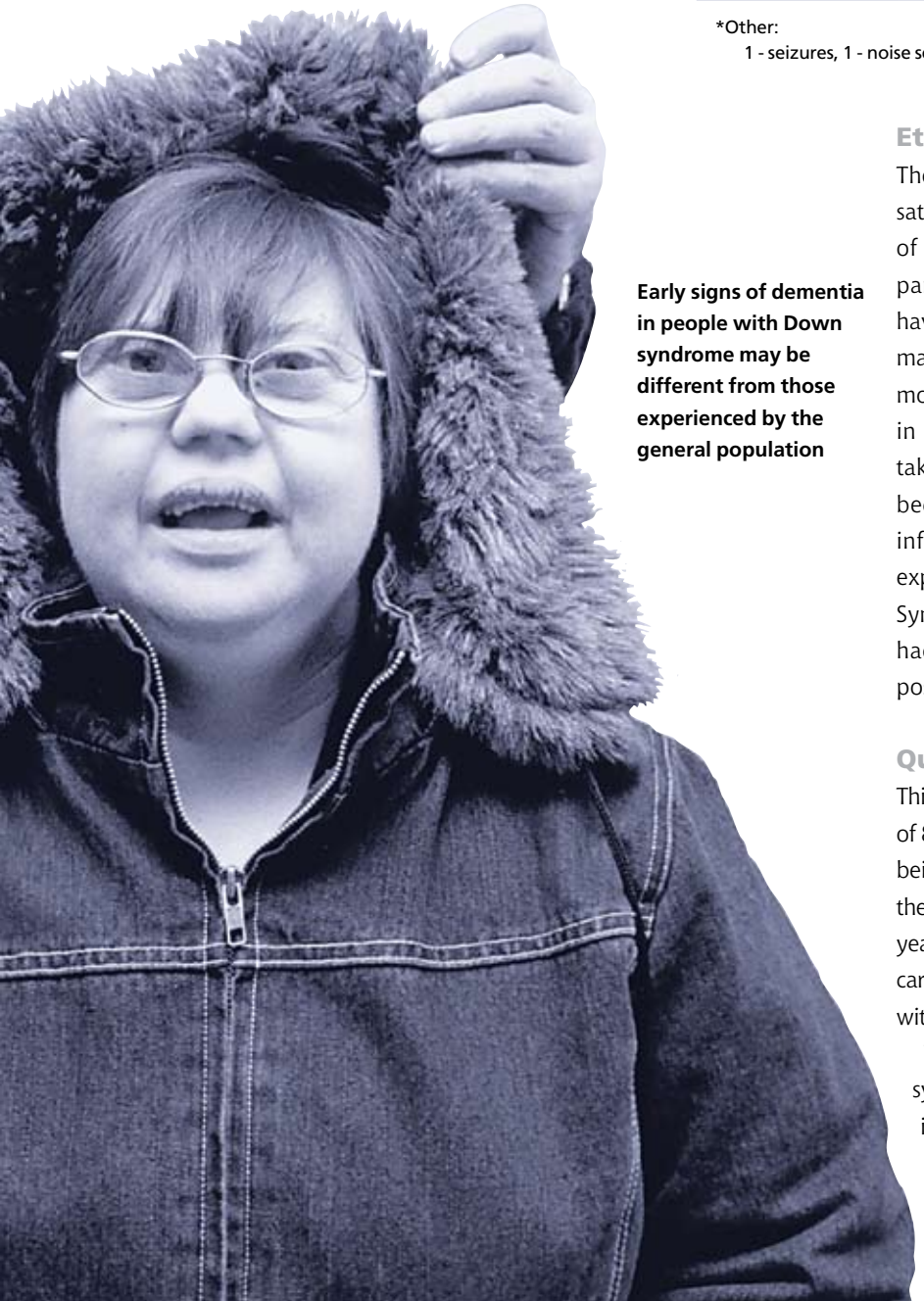
invited to complete a questionnaire. To gather the most up-to-date information in Scotland as possible there was a requirement that participants had been caring for a person with Down syndrome and dementia for up to five years since diagnosis.

The postal questionnaire was subsequently sent to 45 carer members of Down's Syndrome Scotland with the aim of discovering the early symptoms of dementia in people with Down syndrome as identified by their carer. They were also asked which professionals they first approached with their concerns. Additionally the questionnaire asked whether the diagnosis of dementia was shared with the person with Down syndrome. The potential implications for professionals working with people with Down syndrome before and after a diagnosis of dementia are discussed below.

Table 1 Early signs of dementia noted by carers

Early sign	Noted by family carer n=12	Noted by paid carer n=23	Total n=35	Percentage of group
Change in behaviour	9	19	28	80
Change in living skills	7	18	25	71
Confusion	6	17	23	66
Change in Sleep pattern	4	7	11	31
Change in communication	5	6	11	31
Memory loss	3	7	10	29
Change in understanding environment	2	3	5	14
Other*	1	3	4	11
Change in physical health	1	2	3	9

*Other:
1 - seizures, 1 - noise sensitivity, 1 - talking to dead relatives, 1 - fear of going outside



Early signs of dementia in people with Down syndrome may be different from those experienced by the general population

Ethics

The research was carried out with members of the organisation in which the researcher is employed. An advantage of this was that it gave direct access to many prospective participants, both formal and informal carers. This may have influenced some people's decision to take part. Some may have preferred not to for fear of not remaining anonymous, particularly if they had met the researcher previously in her professional capacity. Others may have chosen to take part because they knew the researcher, rather than because they wanted to be involved in the research. An information sheet made clear the boundaries, roles and expectations. Information was not sought about Down's Syndrome Scotland, the organisation through which access had been obtained, as this may have compromised the position of the participants.

Questionnaire findings

Thirty-five questionnaires were returned, giving a response rate of 80 per cent. The average age of those with Down syndrome being cared for by participants was 52.8 years at the time of the diagnosis of dementia. The average age of men was 51.4 years and of women, 53.9 years. The average age of family carers was 79.5 years for parents and 51.3 years for siblings, with the average age of paid carers being 42.5 years.

The first signs of dementia in the person with Down syndrome noted by their carers were a change in behaviour (80 per cent), change in living skills (71 per cent) and confusion (66 per cent), as detailed in Table 1. This

Table 2 First person spoken to by the carer

First spoken to	Family carer	Paid carer	Total	Percentage of group
General practitioner	5	12	17	49
Community learning disability nurse	1	4	5	14
Family member	2	1	3	9
Day care staff	1	2	3	8
Colleague	0	2	2	6
Voluntary organisation	2	0	2	6
Psychiatrist	0	1	1	3
Social worker	1	0	1	3
Psychologist	0	1	1	3
Occupational therapist	0	0	0	0
Speech and language therapist	0	0	0	0
Person with Down syndrome	0	0	0	0
Advocate	0	0	0	0

was followed by a change in sleep pattern (31 per cent), communication difficulties (31 per cent) and memory loss (29 per cent). Lesser noted signs were a change in understanding the living environment (14 per cent) and a change in physical health (9 per cent).

Carers most commonly shared their first concerns with the general practitioner (GP) for the person with Down syndrome (49 per cent), as shown in Table 2. Other first points of contact for carers were community learning disability nurses (14 per cent), family (9 per cent), and day-care staff (8 per cent).

Tables 3 and 4 show that the diagnosis of dementia using the word 'dementia' or one of its forms such as 'Alzheimer' was shared with the person with Down syndrome in just four cases (11 per cent). Carers used words other than 'dementia' or 'Alzheimer' in five instances (14 per cent). Twenty-seven of the 35 people with Down syndrome (77 per cent) cared for as part of the research were not told anything at all about their diagnosis. Seven of the carers gave as a reason their belief that the person they cared for would not understand the diagnosis.

Discussion

Age at diagnosis

The average age of people with Down syndrome of 52.8 years when diagnosed with dementia is younger than the findings of Tyrell *et al* (2001), who recorded it to be 54.7 years. The average age of family carers who cared for a person with Down syndrome and dementia at 79.5 years is older than family carers of people with dementia in the wider population, whose average age was recorded as 72.1 years (Ballard *et al* 1996). This can be attributed to carers in Ballard's study generally caring for a spouse or older parent, whereas participants in this research were caring for an adult child.

The average age of siblings at 51.3 years is likely to coincide with additional responsibilities for older children or grandchildren, ageing parents or their own health problems (Fray 2000). Issues for service planners and care providers here include the additional stress experienced by carers of all ages.

Early indicators of dementia noted by carers

The lack of reports of memory loss as a first indicator of dementia in people with Down syndrome echoes findings from elsewhere (Ball *et al* 2006, Kittler *et al* 2006). The first issue this raises is that if the dominant early signs of change in behaviour, confusion and change in living skills are not recognised as being possible early indicators of dementia, then a potentially very important diagnosis may be missed and not made until much later. The implications of this range from losing the opportunity to prescribe medication, if appropriate, in the early stages to unnecessary delays in planning for the person's future life with the condition. Keeping dementia in mind as just one possible reason for the changes would also entail a search for other health issues that may exacerbate the situation, such as sensory losses, thyroid problems, mobility changes and bowel dysfunction, all of which are common in people with Down syndrome.

A further issue is that although the majority of people with Down syndrome are known to be at risk of Alzheimer's disease (Devenny *et al* 2000), this is not the only form of dementia that occurs in this population. Instances of Lewy body, small spots in damaged nerve cells at the top of the brain (Simard and van Reekum 2004), and frontotemporal dementia (Ball *et al* 2006) have been recorded. Early symptoms and signs are consistent with those seen in the general population and may differ not only between the different forms of dementia but also from early features of Alzheimer's type dementia. For example, early symptoms in patients with frontotemporal dementia include difficulties in communication and a change in personality (Diehl-Schmid *et al* 2006). Loss of concentration, changes in behaviour and hallucinations have been recorded as prominent early symptoms

in dementia with Lewy bodies, whereas a loss of short-term memory is predominant in early stage Alzheimer's type dementia (Ferman *et al* 2006).

Further research is needed to look for effects of different underlying pathologies when several types of dementia co-exist in people with Down syndrome. Understanding this is crucial to determining the diagnosis, interventions and support for each individual, just as it is in the general population.

First contact for concerned carers

Unsurprisingly, with relatively easy access to primary care services, the most common first contact for carers was the GP of the person with Down syndrome. What the GP subsequently does, and who else becomes involved in future planning, is vital in ensuring that social and environmental issues are considered, in addition to physical health.

Bond *et al* (2002) referred to a medicalisation of dementia, with GPs being viewed as experts who make a diagnosis and recommend treatment, yet do not always refer to community-based or non-medical professionals who may be able to offer different supports or interventions. Despite the strong emphasis placed on GPs by carers there is a lack of training available for them in Down syndrome and dementia (Kerr *et al* 2006). In Scotland a GP is likely to have no more than five people with Down syndrome registered at any time on his or her practice list, making this group numerically a low priority compared to others with dominant health issues (NHS Health Scotland 2004).

Nine per cent of carers first contacted staff at the day service used by their family member to discuss specific concerns. In all cases this was a learning disability-specific environment. Day care facilities for people with dementia in the general population have been seen to be valuable and supportive environments (Downs *et al* 2002). For people with Down syndrome it is often one of the first parts of their daily routine that stops or is reduced. This is due to perceived difficulties anticipated by staff, inappropriate noisy environments, activities it is thought the person no longer enjoys or difficulties with travel arrangements (Whitehouse *et al* 2000). This means that a familiar service is no longer available. Another consequence is that older family carers who previously had a period of regular respite are left with additional daytime responsibilities.

Sharing the diagnosis

Research by Pinner and Bouman (2003) found that 98 per cent of carers wanted to be told should they be diagnosed with dementia in the future. But this question is still not being asked of people with Down syndrome. People are not asked if they would like to know what might be happening to them when they experience changes or whether they would like to be told under similar circumstances in the future (Stalker *et al* 1999). One assumption is that they would not have the ability to understand their diagnosis. This is despite research findings that suggest that non-disclosure of diagnosis in the general population can have a negative impact on the person and their family (Fearnley *et al* 1997).

Pratt and Wilkinson (2003) suggested a psychosocial model for people in the general population who have dementia, which may also be appropriate for people with Down syndrome. This demonstrates an awareness that an unsupportive environment can be created if a person wants to know what is happening to them but is denied a diagnosis.

If carers do not know if or how a diagnosis has been given, either explicitly using the correct terms or implicitly using words such as 'confused' or 'not well', they will not be able to discuss current or future plans with the person with Down syndrome. This can lead to a lack of forward planning for the later stages of dementia and may increase the sense of isolation and frustration among people with Down syndrome. This is consistent with a reactive rather than proactive approach to health care, as seen in Scotland (NHS Health Scotland 2004). The emphasis is on an individual seeking help rather than support being offered.

Table 3 Client's awareness of dementia diagnosis

Given diagnosis	Supported by family carer	Supported by paid carer	Total	Percentage of group
Yes	1	3	4	11
No	11	20	31	89

Table 4 Client's awareness of memory problems

Told of changes	Supported by family carer	Supported by paid carer	Total	Percentage of group
Yes	4	0	4	11
No	7	16	23	66
Already told had dementia	1	3	4	11
Not answered	0	4	4	11

Recommendations for practice

- Increased awareness and training for carers and professionals, including GPs, on the early signs of dementia in people with Down syndrome. This should include different types of dementia and highlight the need for an integrated approach to the planning of subsequent care. The emphasis should be on co-ordinated medical and social care rather than reliance on a sole discipline.
- One named individual should take responsibility for explaining to the person with Down syndrome why he or she is experiencing the changes. For consistency, the signs or words used should be recorded and shared with all those involved in the person's care. This will enable future discussions to include the person with Down syndrome.
- Disclosure of a diagnosis should be seen as a starting point for future support, not an end result. This means that care should be planned from that point onwards to anticipate changing needs, including appropriate daytime activities.

The postal questionnaire from Down's Syndrome Scotland showed that 77 per cent of the study population were not given any information about the changes that they were experiencing. There seems to be no reason why this should not lead to the same confusion, distress and agitation as has already been noted in the general population (Bamford *et al* 2004). Diagnosis would give valuable time in the early stages for the person with Down syndrome to make decisions about their future and to allow for carers to access support and information for themselves and the person that they care for.

Early signs

Early signs of dementia in people with Down syndrome may be different from those experienced by the general population, and a failure to recognise this can lead to a delay in diagnosis and subsequent interventions.

Consideration should be given to the reported early signs being experienced, which may help with an accurate diagnosis of the specific type of dementia. It should not always be assumed that the person will have Alzheimer's disease. Other forms of dementia, with different early signs, can occur. Crucial to an integrated care plan is the involvement of health, social care and community-based professionals in addition to the GP, although it is currently the GP who carers usually seek help from first.

The diagnosis of dementia, either explicitly or implicitly, is not being given to people with Down syndrome. This means that current and future choices over interventions, health care, day services, accommodation and ultimately end-of-life care are not being offered. Giving a diagnosis in an appropriate way should be seen as the start of the process of supporting people through the disease ■

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